

Propionic Acidemia (PA)

An organic acid disorder

What is it?

Propionic Acidemia (also known as PA) is an inherited organic acid disorder. People with organic acid disorders, like PA, cannot properly break down certain components of protein and fats. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down the proteins and fats, certain organic acids build up in the blood and urine and cause problems when a person eats normal amounts of protein, or becomes sick.

What are the symptoms?

A person with PA can appear normal at birth. Some people with PA will have the following symptoms after a few days of life: poor feeding, lack of energy, vomiting, low muscle tone, and seizures. There is a high risk of infection, developmental delay, seizures, and neurological problems. Many symptoms of PA can be prevented by immediate treatment and lifelong management. People with PA typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

PA is inherited in an autosomal recessive manner. This means that for a person to be affected with PA, he or she must have inherited two non-working copies of the gene responsible for causing PA. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have PA. Typically, there is no family history of PA in an affected person. PA is a rare fatty acid oxidation disorder; about 1 in 100,000 babies born have PA.

How is it detected?

PA can be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

PA is treated by eating a diet low in protein and drinking a special formula, and sometimes medication, as recommended by a genetic metabolic medical professional.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: <http://ghr.nlm.nih.gov/ghr/page/Home>

Save Babies Through Screening Foundation

4 Manor View Circle Malvern, PA 19355-1622 Toll Free Phone: 1-888-454-3383

Fax: (610) 993-0545 Email: email@savebabies.org

Website: <http://www.savebabies.org/diseasedescriptions.php/>

Organic Acidemia Association

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Propionic Acidemia Foundation

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Highland Park, IL 60035

1-877-720-2192

www.pafoundation.com □

STAR-G Hawaii Department of Health

<http://www.newbornscreening.info/Parents/organicaciddisorders/PA.html>

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